Identifying Medically Relevant Variation to Re-Classify Disease: Linkage Analysis of Neurodegenerative Disorders in the 1980's

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Abstract

Mapping technologies have been widely used in evolutionary biology and biological anthropology, but also human geneticists have relied on them to identify hereditary modes of disease contagion, and to correlate diseases like haemophilia with other biological properties such as male sex. The increased use of molecular methods in the second half of the last century, e.g. the identification of restriction fragment length polymorphisms, lead to new hopes to successfully use linkage analysis in research on seemingly hereditary neurodegenerative disorders such as Huntington's disease or Alzheimer's disease.
In my talk, I want to take a closer look at the ways in which the disease definitions of these neurodegenerative disorders have been amended so as to provide suitable objects of analysis, and, in turn, how the results of linkage analysis research have stabilised or de-stabilised existing disease classifications. Classifications are understood as definitions of disease, which allow for ordering individual disease phenomena into groups according to a selection of classifiable characteristics.
My talk addresses such selections by analysing how experimental practices of linkage analysis and conceptual work regarding the search for good classifications of the named neurodegenerative disorders were intertwined, that is to say, how the diseases were treated as both, markers and yet-to-be-ordered-phenomena. Against this background, it shall be discussed how human geneticists dealt with the heterogeneity of disease phenomena, for instance by sub-typing patient populations, by re-framing clinical hallmarks, and by negotiating which individuating properties could be disregarded for the sake of building general, medically relevant categories.

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